MULTI-DISIPLINARY TEAM CARE GUIDELINES

1302 Watson Blvd #1015, Warner Robins, GA 31093 USA
www.curevcp.org
https://flow.page/curevcpdisease
info@curevcp.org

CONNECT WITH OTHER FAMILIES

DIAGNOSIS
A mutation in the VCP gene causes multisystem proteinopathy 1 (MSP1). Genetic testing remains the only definitive way to diagnose this condition. The VCP protein has a wide variety of functions within cells, and a variety of conditions may occur in an individual when a VCP gene variant is present.

CARE GUIDELINES
Scan for publication in the Orphanet journal of rare diseases

ABOUT THE DISEASE FOR PATIENTS AND FAMILIES

FEATURES
This genetic disease can affect the muscles, bones, nerves, and brain. Individuals with this condition typically do not develop symptoms until mid-adulthood and may only exhibit one symptom.

Symptoms vary from person to person, even among family members. It is unknown how many people are affected with this condition in the world, but it is extremely rare. Even though disease-modifying therapies do not exist for many of the conditions, interventions and supportive therapies can help improve quality of life.

Multi-disciplinary care is vital to screen and treat the various symptoms that may develop over a person’s lifetime. Work with your team of doctors and therapists to develop an individualized, comprehensive care plan.

INHERITANCE
This condition is inherited in an autosomal dominant pattern, which means that an affected individual has a 50% chance of passing the VCP mutation along to a child.

MULTI-DISIPLINARY TEAM
The care team may include:
(depending on your conditions)

- Neurologist
- Endocrinologist
- Psychologist
- Pulmonologist
- Cardiologist
- Geneticist
- General Practitioner
- Physical Therapist
- Occupational Therapist
- Speech Language Pathologist
- Respiratory Therapist
- Genetic Counselor
- Social Worker
- Caregiver/Family Support

POTENTIAL SYMPTOMS
Tell your medical provider if you are experiencing

These symptoms:
- Pain
- Weakness
- Muscle loss
- Muscle cramps, spasms, twitches, or tremors
- Tingling in hands or feet
- Recent falls
- Bone fractures or deformities
- Hearing problems
- Trouble swallowing
- Shortness of breath
- Heart problems
- Fatigue

Difficulty with:
- Walking or climbing stairs
- Standing up
- Lifting and carrying heavy things
- Housework and yardwork
- Handwriting or typing
- Toiletting and hygiene
- Frequent urgency to use the bathroom
- Eating
- Sleeping
- Communication
- Engaging in social activities
- Change in behavior or mood

In previous days, the condition was known as: Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia (IBMPFD)

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JOINT THE PATIENT REGISTRY
Your participation helps advance therapeutic development.
www.curevcp.org/patient-registry
CLINICAL MANIFESTATIONS OF VALOSIN-CONTAINING PROTEIN (VCP) ASSOCIATED MULTISYSTEM PROTEINOPATHY (MSP)

关于疾病的医生和提供者

- 轴向和近端无力，进展至远端，最常见。
- 出现面部、肩部、手臂肌肉无力、上眼睑肌肉无力、下眼睑肌肉无力，描述为肢带肌无力症、眼咽部肌肉无力症、远端肌无力症。
- 骨痛、骨变形、病理骨折、听力损失。
- 快速进展的行为障碍、执行功能障碍、语言障碍。
- 常常伴有帕金森表现，如震颤、僵直、步态障碍。
- 反复呼吸道感染、咳嗽无力、吞咽困难、睡眠呼吸暂停、呼吸衰竭。
- 多焦点无力、高反射或低反射、萎缩、颤搐、咀嚼无力、呼吸肌涉及、体重损失。
- 基础运动障碍、自主功能障碍、各种非运动性症状。
- 记忆力障碍，以记忆性障碍和高阶认知功能障碍为主。
- 长度依赖性无力，高反射性，痉挛。
- 长度依赖性无力、肌肉萎缩和感觉障碍。
- 营养脚变化和远端无反射。
- 努力性呼吸困难，心力衰竭。
- 吞咽功能障碍，讲话音量和理解力降低。